**Copy number of human SULT1A1 gene among newly menopausal Caucasian women**

Ann Moyer, Richard M Weinshilboum, Virginia M Miller. Molecular Pharmacology and Experimental Therapeutics, Surgery, Physiology and Biomedical Engineering, Mayo Clinic College of Medicine, Rochester, MN

There is large variation in response to menopausal hormone treatments among women and it remains unclear as to what genetic factors might increase risk of breast cancer in women using such treatments to relieve symptoms of menopause. SULT1A1 is a ubiquitous enzyme involved in the metabolism of steroid hormones including estrogen. A number of polymorphisms in the gene for this enzyme are associated with various cancer risks and the activity level of the enzyme is correlated closely with variation in copy number of the gene. This pilot study evaluated SNP variation and copy number of SULT1A1 in a subset of women (n=156) being screened for the Kronos Early Estrogen Prevention Study (KEEPS). All participants gave written informed consent. DNA prepared from blood was genotyped for SULT1A1 copy number, rs3760091 and rs750155 from the promoter region of SULT1A1, and rs9282861 from the coding region (SULT1A1*2 variant). In this cohort, 5.2% had 1 copy of SULT1A1, 65.2% had 2 copies, 23.9% had 3 copies, and 5.8% had 4 copies. Previously reports of the allele frequency for the three SNPs did not take into account gene copy number. When copy number was taken into account, allele frequency was significantly different, a fact that will have to be taken into account in future genotype-phenotype correlations.